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A Case Revisited: Recent Presentation of Incontinentia Pigmenti in Association With a Previously Reported X;Autosome Translocation

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INTRODUCTION

Incontinentia pigmenti (IP) comprises unusual pigmentation and malformation of eyes, teeth, skeleton and heart. Swirling areas of hyperpigmentation follow the lines of Blashko. The first (vesicular) stage of the skin lesions may occur a few days after birth and consists of a linear array of erythema and bullae on the limbs; 95% of patients will have such lesions by the age of 1 year. The second (verrucous) stage generally occurs after the vesicular lesions have regressed and is characterized by pustules, papules and hyperkeratotic streaks. Stage 2 lesions are found in 70% of patients with IP. Stage 3 (pigmented) is characterized by hyperpigmented whorled macules. Stage 4 consists of the fading of hyperpigmented lesions and the development of atrophic scars. IP is an X-linked dominant condition with lethality in males. The phenotype of IP in affected females is consistent with random X inactivation. The gene for IP1 was localized to Xp11 as the result of identifying individuals with translocations involving this region [Hodgson et al., 1985; Gilgenkrantz et al., 1985; de Grouchy et al., 1985; Kajii et al., 1985; Cannizzaro and Hecht, 1987; Crolla et al., 1989; Bitoun et al., 1992].

We reported 2 unique cases with de novo unbalanced mosaic cell lines in which each cell line had a different structural abnormality involving the same region [Pettenati et al., 1993]. The second case involved a 3-day-old black girl referred for cytogenetic testing because of congenital anomalies. Her karyotype was interpreted as 46,X,idic(Xq)/46,X,idic(Xq),der(12)t(X;12)(p11.2;p13.3). Parental chromosomes were normal. At 7 months, she was small for her age, had a depressed nasal bridge, marked epicanthic folds, a thin upper lip vermilion, mild micrognathia, tongue protrusion and a rather "coarse" facial appearance. She had global developmental delay. She was lost to follow-up, but recently was re-referred at $4\frac{7}{12}$ years for reevaluation. She now has skin findings consistent with classic IP stage 3. There were swirling areas of alternating hyper/hypopigmentation over the back, trunk and limbs (Fig. 1). A review of her karyotype shows interruption of the Xp11.2 region that is associated with IP. This case then represents another confirmation of the association of IP with Xp11. Her length was 91.5 cm (mean height age for 2.5 years), weight 10.6 kg (mean for 15 months), and her head circumference 44.5 cm (mean for 10 months). She has alternating exotropia. The right upper central and lateral incisors were fused, the remaining teeth being widely spaced. Her fingers were tapered with finger-like thumbs. In addition, there were mild flexion contractures of the third and fourth fingers bilaterally at the PIP joints. There was generalized hypotonia.

This case underscores the need for follow-up of patients with chromosome abnormalities. Despite the disruption of the Xp11 band in this patient, she did not exhibit the typical IP at birth and due to the lack of routine follow-up, we do not know when the IP signs first developed. This case represents another confirmation of the association of IP with Xp11.

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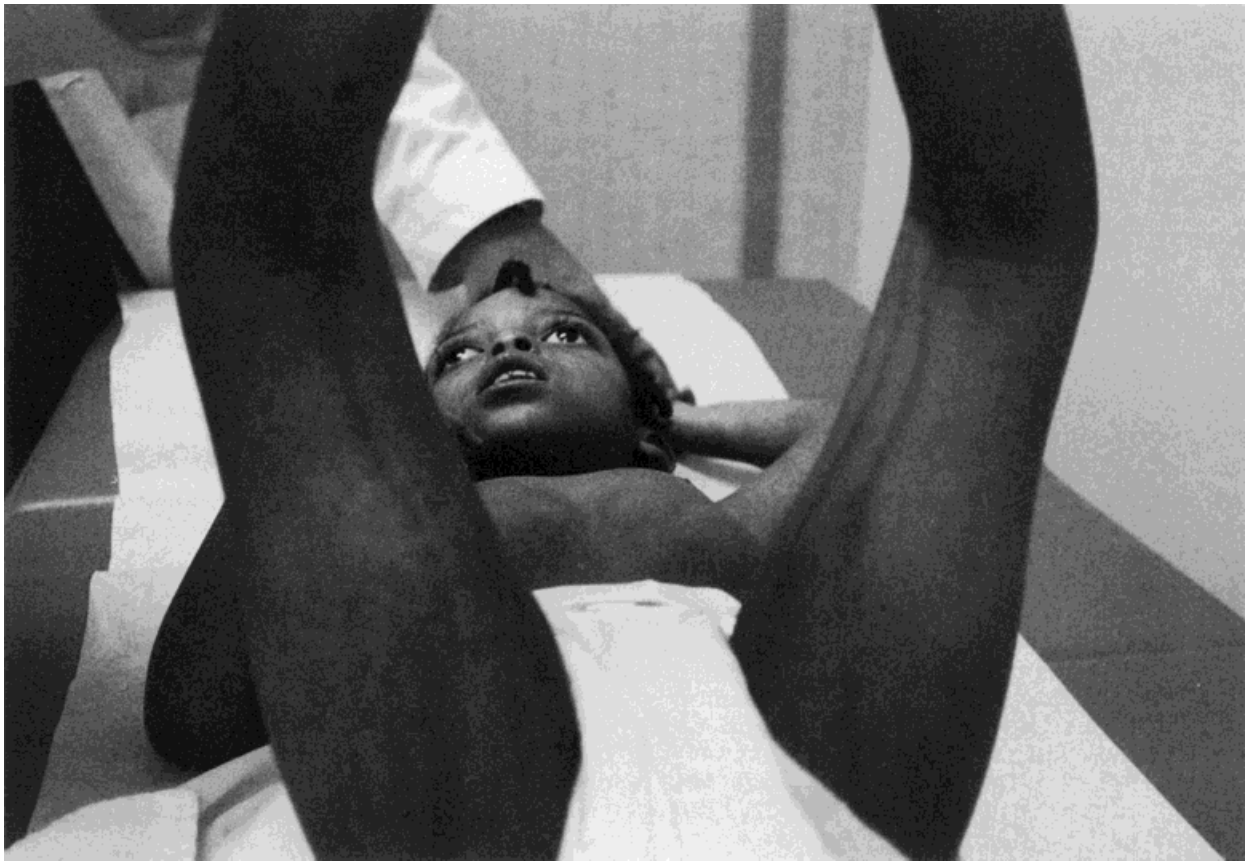

Fig. 1. Photograph of proband at 4 years, 7 months of age showing streaky hyper/hypopigmentation on the legs.

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